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ABSTRACT

This pamphlet reviews the historical process involved in initially recognizing Rett Syndrome as a specific disorder in girls. Its etiology is unknown, but studies have considered factors as hyperammonemia, a two-step mutation, a fragile X chromosome, metabolic disorder, environmental causation, dopamine deficiency, and an inactive X chromosome. Characteristics of Rett Syndrome include onset during the second year of life, acquired microcephaly, stereotypical hand movements, ataxia, and a consequent classification as severely or profoundly retarded. Girls with Rett Syndrome have secondary problems such as scoliosis, seizures, respiratory problems, teeth-grinding, communication disorders, and poor motor skills. Some medications have been effective in controlling the seizures, such as "Tegretol," "Depakene," "Dilantin," and "Clonopin-Clonazepam." Special teaching techniques consist of music therapy, swimming therapy, tactile stimulation, horseback riding, etc. Adapted classroom equipment appropriate for use with Rett Syndrome children includes scoliosis jackets, corner chairs and other adapted chairs, bolster rolls, vestibular boards, large balls, and wheelchairs. (JDD)

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RETT SYNDROME

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Rett syndrome is a disorder that affects only girls. It was first described by Dr. Andreas Rett in Vienna Austria in 1966. Dr. Rett did much of his work in institutions working with the mentally retarded. One day, he noticed in his waiting room two women sitting side by side, each with a little girl on her knee. The children had strikingly similar appearances, and both were performing complex wringing motions with their hands. This prompted Dr. Rett to re-examine his files, and he found twenty-two girls who all displayed the same stereotypic movements. He spent a considerable time researching in an effort to identify the specific disorder from which they were suffering. At this point he did not realize that he had identified an entity which had not been previously described. Because his research did not produce an answer, he made a videotape of the girls and travelled to other centers in Europe showing it to other doctors who treated the mentally retarded. They all stated that they remembered seeing a girl like this at sometime in their practice but could not identify the disorder. Dr. Rett decided to publish his findings, and the first cases of what is now called Rett syndrome were described. The next twenty years were very frustrating for Dr. Rett since his work received very little publicity and little research was done in an attempt to establish the cause of the disorder. The turning point seemed to be the publication of an article in the Annals of Neurology, in October 1983, by Hagberg et al., in which 35 further cases of the disorder were described. This was the first report of the disorder to appear in the English language.

The number of cases of Rett syndrome being diagnosed in the United States and throughout the world is growing very quickly. At the Rett syndrome seminar held in January of 1985, fifty cases had been identified in the U.S. and by November, 1985 this figure had risen to almost 200 cases. The estimated number of cases in the United States is approximately 7,500. This figure is based on the Scottish and Swedish estimated incidences of 1 in 30,000 or 1 in 15,00 live female births. However, because of racial and ethnic differences between American and European populations, it should be borne in mind that the incidence in the U.S. may differ from that found in other parts of the world.

As the stages of the disorder unfold it will be realized what a devastating condition this is for the family of an affected child. The early autistic-like phase puts great strain on the family. In some cases these behaviors appear quite quickly, while in other cases they may take several months to evolve.

On November 20 and 21, 1985, 116 scientists from many countries gathered at the Kennedy Institute in Baltimore, Maryland, to attend a workshop devoted to the possible causes of Rett syndrome. Many suggestions emerged from this workshop, and the papers presented at the meeting have been published in a special issue of the American Journal of Medical Genetics, Supplement 1, 1986.

ETIOLOGY

The etiology of Rett syndrome is unknown. Because of the small number of people afflicted by this syndrome, it is very difficult to compile data and do research into the cause. So far there is no concrete evidence, but it is strongly felt by the medical profession that this is a genetic disorder. It has been reported on all continents, in all major religious and ethnic groups, and in all social strata.

Hyperammonemia

Dr. Rett originally felt from the studies he made that hyperammonemia was a feature of the disorder, but as more cases were identified, this was later disproved. He found that only four out of fifty-four case studies had unusually high levels of ammonia in their blood or urine.

Two-Step Mutation

Dr. Wahlstrom from Goteborg, Sweden, has suggested that a two-step mutation may be the cause. The theory is based on the fact that the condition only affects girls and that cases seem to appear spontaneously in families. His theory contends that there is an autosomal recessive mutation on a somatic cell which in rare cases may be accompanied by a second somatic X chromosomal mutation. Dr. Wahlstrom has done chromosomal studies on fifteen cases and found a high incidence of fragile sites on the X chromosome. He is presently doing DNA studies on the girls and their families but has not seen any marked difference between affected and unaffected girls.

Fragile X

The fragile X theory initially had some support. It could have explained why this syndrome is only found in girls, as affected male fetuses would be lost as spontaneous abortions. Despite extensive studies in a number of laboratories, no consistent fragile site has been identified, and it is now felt that the cause of the disorder is not related to fragile sites on the X chromosome.

Metabolic Disorder

Dr. Rett has described one autopsy done on a Rett girl as having mild, diffuse cortical atrophy and non-specific cell changes (Holm, 1985). Neuropathological studies completed on five other girls who have died showed conditions similar to those seen in aminoacidopathies. This is suggestive of a possible metabolic etiology, but requires much more evidence before this can be proved.

Environmental

References to this disorder date back only twenty years. This does not, however, mean that it did not exist before 1966, but merely that it went unrecognized before that time. While environmental causes are unlikely to be the answer, because of the widespread occurence of the disorder they cannot be excluded. It is possible that a virus or some other agent which would affect the mother during pregnancy but is undetected by her may be responsible. It should be remembered that the Rubella syndrome masqueraded as a genetic disorder for many years before its viral etiology was discovered.

Dopamine Deficiency

Dr. Alan Percy of Baylor College of Medicine has found a reduction in the levels of neurotransmitter metabolites in the cerebrospinal fluid of girls with Rett syndrome. Homovanillic acid, a dopamine metabolite, was reduced to the greatest extent. Whether the reduction in neurotransmitter metabolites is secondary to an abnormality in presynaptic synthesis, receptor sensitivity, or to a defect in the feedback mechanisms, remains to be determined.

Inactive X Theory

Dr. Vincent Riccardi described the inactive X theory at the November 1985 Rett Syndrome Conference. He stated that although girls have two X chromosomes, only one of them is used for metabolic information, the other remaining dormant or inactive until very late in mitosis, when a protein activates it for replication purposes. Dr. Riccardi felt that the protein necessary for this operation might be impaired or missing in a girl with Rett syndrome. He is researching this theory at the present time.

CHARACTERISTICS OF RETT SYNDROME

ONSET

The majority of mothers reported a normal pregnancy, followed by normal perinatal and neonatal periods. Development during the first seven to eighteen months is unremarkable, but then stagnates and a period of rapid deterioration of both behavior and mental ability follows. Some of the parents have described their daughters as seeming to be hallucinating. Some initially go through crying and screaming spells that continue intermittently over several days at a time. Often the girls lose their sleep patterns, become very withdrawn and cease all verbal communication. Most cases during this early onset period are identified as having "early infantile autism" or "autistic-like" conditions or "disintegrative childhood psychosis."

MICROCEPHALY

The microcephaly found in girls with Rett syndrome is acquired, i.e., it is not present at birth. The girls are born with head circumferences which fall into the normal range, but with the onset of the syndrome, head growth slows down. Between the ages of two and four years, the microcephaly is usually noted, although in some children whose head circumference at birth is larger, the microcephaly may not be noted until some years later.

PURPOSEFUL HAND USE

The most obvious characteristic of a girl with Rett syndrome is the stereotypical hand movements. Most girls have acquired their basic hand

skills including the pincer grasp before the onset of the symptoms. Purposeful hand movements are lost by a majority of the girls before the age of four years. Purposeful movements are replaced by continuous stereotypic hand movements. Classically, these are the clasping and wringing of the hands, which have been described as "hand washing" movements and which are usually performed in the midline, close to the chest. Tapping at the mouth, biting or sucking of the hands, and pulling at the hair or ears is also seen. The child is unable to reach or grasp even for the most wanted items. Where grasping does occur it is only momentary, and the item is quickly dropped. Methods to restrain the hands have been tried without great success. Some of the girls seem to become more anxious and nervous and wring even harder when the restraints are removed. Dr. Rett recommended at the conference to not interfere with the hand movements in a continuous effort.

ATAXIA

The average age of walking is twenty-five months, although a small proportion of girls never acquire this ability. The girls who have a late onset of symptoms seem to walk earliest and maintain this ability for the longest time. They demonstrate a jerky truncal ataxia which usually involves the limbs. Some girls demonstrate extreme fear of physical movement, and this adds to the ataxia. Tremors occur in many of the girls and myoclonic movements are also fairly common. The limbs become most ataxic when the girls are trying to maintain their balance or when they are in a disturbed emotional state. Loss of balance is very common, and it is difficult for them to break the wringing motions of their hands in order that they can make use of protective reflexes.

LATER YEARS

By the time the girls reach the second decade of their lives, the regression seems to have levelled off. They are now usually classified as severly or profoundly retarded. Many no longer walk and have poor range of movement with no purposeful hand activity. Personality is usually placid with good eye contact and some vocalization but no meaningful language. Life expectancy depends on the severity of involvement and the quality of the care. It can span many decades. The girls are usually warm and loving, and while they have difficulty expressing their emotions, their loving eyes and warm smiles express more than words could ever say.

ASSOCIATED MEDICAL PROBLEMS AND SECONDARY LEARNING DISABILITIES

Girls with Rett syndrome have a variety of secondary problems stemming from their primary neural deficits. Poor ambulation, scoliosis and contractures are their major problems, and these predispose them to chest infections and pressure sores which must be treated promptly.

SCOLIOSIS

Scoliosis occurs in a high proportion of cases. The onset of the curvature of the spine is usually at the end of the first decade but can begin earlier. It usually becomes more severe after the loss of ambulation. Preventive measures should be taken long before the scoliosis appears. These measures should include good positioning at all times and frequent changes of position, using a wide variety of positions for both sitting and lying. A good physical therapy program is essential, with use of many range of motion exercises. Many times the child may appear to have a scoliosis before the spine actually develops a curve. A trunk jacket can be used as early as possible in an attempt to prevent further damage, if the child's orthopedist feels it would be of benefit. Frequent spinal x-rays and physical examinations should be made.

SEIZURES

The electroencephalogram (EEG) is usually abnormal by three years of age. It is estimated that a majority of girls with Rett syndrome suffer from seizures. The abnormality seems to be predominantly in the left temporal, parietal, and frontal lobes. The most common type of seizure is the tonic-clonic variety. The sleep patterns of the girls are very erratic, especially during the regression phase of the disorder. Much of the seizure activity seems to occur during sleep. The seizures can be controlled by the use of conventional drugs, although seizure management is difficult in a small minority of the girls. The most effective drug seems to be Tegretol, but again, a small number of children require other drugs, or even combinations of drugs. Seizures can cause a dramatic change in behavior and can also make the child extremely tired with a need to sleep for a period of time. A great deal of energy can be expended during a seizure, and professionals who deal with Rett syndrome girls need to be very much in tune with their behavior patterns so that they can recognize when to push them and when to allow them to relax.

RESPIRATORY PROBLEMS

Due to the variable muscle tone in the chest wall and to the scoliosis, many of the girls suffer from repeated respiratory problems. When they get an infection they cannot always coordinate their coughing which would help to clear the mucus in their lungs. Many are mouth breathers and their nasal passages may remain congested for longer than is usual with such infections. Lung congestion can lead to bronchitis and pneumonia. This may cause them to miss a great deal of school. During periods of infection, positioning is very important. The child should not be laid supine, as this position does not facilitate good coughing. The best position would be on the stomach with the chest lower than the abdomen to encourage drainage; or, with the head slightly elevated to allow for more restful sleep. Conventional decongestants can be used but may have side effects such as drowsiness, and this may depress

the ability to cough. Be cautious of using proprietary medicines if the child is taking any other medications, especially those for seizure control. A medical doctor should be consulted before combining medications.

TEETH-GRINDING (BRUXISM)

A significant proportion of the girls are teeth-grinders. They display this behavior in varying degrees. Many are not severe enough to require a dental guard, but a minority would benefit from a suitable appliance. Some research has been done to develop a suitable guard, and currently Ralph B. Sviger, D.D.S., of Leesburg, Virginia, has developed a mouth/jaw plate to help a Rett syndrome girl who has developed a severe jaw misalignment as a result of excessive grinding.

Dental problems can be very painful and can complicate feeding. Correcting problems such as grinding the teeth can be more painful than the condition itself, but may be a necessary step when the problem is severe. A few of the complications that need to be addressed before using an appliance to correct the child's bite or grinding behavior are the following:

a) Will it interfere with the child's feeding to the degree that it could be hazardous to her health?
b) If the child is a mouth breather, could the appliance interfere with breathing, especially in a child with respiratory problems?
c) If the girl has seizures, could the appliance break during a seizure?
d) Is the pain and discomfort involved going to interfere with the child's emotional stability?

COMMUNICATION

All of the above mentioned secondary problems can cause learning disabilities. It seems that the most severe learning disability the girls possess is the lack of communication modalities. These girls lose most, if not all, of their verbal abilities very early in the progression of the syndrome. Their kinesthetic abilities are greatly impaired by the physical handicaps caused by the syndrome. Their hand movements make it impossible for them to use signing and even pointing to a picture is beyond their ability. The most difficult part of teaching these girls is to find a way for them to show you what they need, want and understand. It is very important not to underestimate the girls' mental abilities due to one's own inability to reach them. Cue into the expression in their eyes or small verbal sounds. Also, tension or relaxation in the body can be an excellent transmitter of the child's likes and dislikes.

MOTOR PROBLEMS

Motor problems in the Rett syndrome child cause more than the obvious learning problems. The girls do not have a good appreciation of spatial relationships, do not seem to know where their bodies are in space, and have very poor motor planning skills. Visual motor skills are also impaired. These skills should not be overlooked during the teaching day. Although weak, they should receive attention to facilitate the development to maximum potential. Active intervention will delay the deterioration in physical abilities.

MEDICATIONS

The most common need for the use of medication in girls with Rett syndrome is for seizure control. The following are a few of the conventional drugs used for seizures and some of their side effects:

Tegretol

Tegretol is the most common and effective drug for the control of seizures. The usual dosage of this medication is between 500-1200mg daily. Some of the side effects are dizziness, vertigo, drowsiness, fatigue, ataxia, poor coordination, confusion, agitated depression, headache, visual hallucinations, talkativeness, speech disturbances, and abnormal involuntary movements. The cardiovascular side effects can include congestive cardiac failure, hypertension, hypotension, edema, and aggravation of coronary artery disease. Intestinal side effects may include nausea, vomiting, abdominal pain, diarrhea and constipation.

Depakene

The average dosage of Depakene is between 15-30mg per dose. This drug is used for simple and complex absence seizures. It is often used in combination with other drugs. Side effects include sedation, emotional upset, depression, psychosis, aggression, hyperactivity and behavior deterioration. Nausea, vomiting indigestion, diarrhea, abdominal cramps, constipation, increased appetite, weight gain and inhibited platelet aggregation can also occur.

Dilantin

Dilantin is most commonly used to control grand mal and psychomotor seizures. The recommended dose in children is 5-7mg per day. Side effects include ataxia, slurred speech, confusion, dizziness, insomnia, nervousness, twitching and headaches. Other side effects include blood disorders, hypotension, conduction depression, ventricular fibrillation, blurred vision, nausea, vomiting, constipation and skin rashes.

Clonopin-Clonazepam

The average dose of Clonopin is between .01 to .03 mg/kg daily. This drug is used to control petit mal, akinetic and myoclonic seizures. Some of the side effects are drowsiness, ataxia, behavioral disturbances, increased salivation, constipation, nausea and skin rashes.

IT SHOULD BE STRESSED THAT SIGNIFICANT SIDE EFFECTS ARE NOT COMMON. BLOOD TESTS TO CHECK THAT THE DRUG IS PRESENT IN THE BLOOD IN THERAPEUTIC AMOUNTS, AND ALSO TO CHECK FOR ADVERSE EFFECTS, ARE USUALLY ARRANGED BY THE PRESCRIBING PHYSICIAN ON A REGULAR BASIS.

EDUCATIONAL PROGRAM PLANNING

Educational program planning for a girl with Rett syndrome must be extremely flexible and change with the day to day condition of the child. The girls need as much one-on-one attention as possible. If the girl is still walking, she may need constant supervision if she is shaky on her feet and falls very easily using little or no protective reflexes. The classroom and home should be made as safe as possible by removing sharp objects and corners which may cause injury.

The earlier programming begins and basic skills are developed, the better the child will do. The program may have to be adapted as the regression progresses. The child should be encouraged to be as independent as possible.

The child's reaction time is usually very slow, so those working with her will need to be patient and allow a great deal of time for each activity. The child should not be frustrated by going on to a new question before she has time to respond to the first. The child's eyes are probably the best indicator of whether or not she is becoming frustrated.

A classroom should have sufficient staffing to meet the needs of a child with Rett syndrome.

Special Teaching Techniques

Special teaching techniques can be as varied and innovative as each teacher. Identified below are some of the more successful teaching techniques used:

Music Therapy

Dr. Rett has noted the benefit of music therapy. The piano seems to be most successful in getting a response. Listening to piano music seems to be calming and reduction of the stereotypic hand movements may be seen. Getting the girls actively involved in playing the piano can help to improve good reaching and hand movements. Other forms of music therapy can also be beneficial, especially those involving instruments which vibrate, i.e., drums, cymbals, etc. Some girls respond very well to "polka" type music.

Swimming Therapy

As with many physically impaired persons, swimming is an excellent form of therapy. It may be possible to do some exercises in water which cannot be accomplished outside the pool. Also, water play is very good for the vestibular system and can provide touch and pressure stimulus over much of the body surface. It is important to ensure that water temperature is sufficiently warm, as many of the children suffer from vasomotor instability which makes the use of a heated pool essential.

Tactile Stimulation

When the child is engaged in programming but is spending time wringing or patting the hands, etc., tactile gloves can be used to provide some tactile stimulation. Provide a variety of gloves with different textures attached to them. This can help keep the child tactilely aware and stimulated. Instead of gloves, materials of different textures can be affixed to tray or table surfaces to provide tactile stimulation.

Horseback Riding

Horseback riding has been found to be beneficial for some Rett syndrome girls. The bouncing motion seems to be very relaxing. It is also excellent for weight shifting and balance. Posture seems to be greatly improved while the girl is on the pony. Since the girl cannot ususally hold on, it is very important to have a placid pony and many side walkers.

Other Teaching Techniques

For many of the girls, to encourage any functional hand use, one of their hands must be held down. This breaks the wringing action and provides less frustration for them.

It is helpful to use a lot of eye contact and facial expressions, which facilitate the personal contact to which the girls are responsive. While working with the child, the teacher should be close to her when talking and direct comments to her.

The teacher should provide a lot of verbal communication and positive reinforcement. The child's eye contact can be used to gauge her response.

ADAPTED CLASSROOM EQUIPMENT

Scoliosis Jacket

Since scoliosis occurs in a high proportion of the cases, a scoliosis or trunk jacket may be needed. It is important to begin using the trunk jacket as soon as any scoliotic signs appear, such as leaning to one side for an extended period of time. Trunk jackets are specially molded for each person and need to be checked frequently for proper fit. Time spent in the trunk jacket needs to be built up slowly and this is usually done in the classroom. During the first days of wearing the jacket, the child may be fussy and uncomfortable. Be prepared with enjoyable activities that can keep her mind occupied during this adjustment period.

Corner Chair

The ability to long sit seems to be one of the first gross motor skills to be lost. A corner chair will facilitate long sitting and keep the pelvis properly positioned. It is very important for the child to spend time long sitting to keep the muscles in the back of the legs extended. Having a corner chair with a tray will make it more functional, i.e., another work station for the child to use throughout the day.

Adapted Chair

It is important to have a properly fitted, comfortable chair for the child to sit in whether she is actively or passively involved in a work setting. The chair should have arm rests that are adjustable. The child should be comfortable but should not rely on the chair totally to hold her up. Head wings will be necessary for the child who has difficulty holding her head up. An adjustable foot rest is also necessary. The child's feet should touch the foot rest. A post between the legs will facilitate good posture and keep the pelvis aligned under the body. Straps may be necessary to keep the child pro-

perly positioned. A tray on this chair would make it extremely functional, serving a variety of purposes.

Bolster Rolls

A variety of positioning and therapy can be done using a bolster roll. It is beneficial for the child with scoliosis to be positioned over the bolster to prevent the progression of the spinal curve. Also, weight bearing activities can be done over a bolster. With the child sitting on the bolster, weight shifting activities can be done.

Vestibular Board

The vestibular board can be used for several activities. It can be used for weight bearing and weight bearing activities. Protective reflex activities can be done on the vestibular board. It is also a good piece of equipment for trunk rotation exercises.

Large Ball

A large ball that is approximately waist high on the girl can be an invaluable piece of equipment. Some of the activities which can be done with the ball are weight bearing on the hands, stomach muscle exercises, weight shifting exercises, bouncing and vestibular exercises.

Wheelchair

As the syndrome progresses, there may be need for a wheelchair. This should be prevented as long as possible, but when the time comes, it is important to have a properly fitted chair that is also functional. The chair should be specially adapted for the individual child. The child should not spend great lengths of time in the chair, and should be repositioned frequently. All pressure points should be checked regularly for signs of tissue breakdown.

Other Equipment

Although the characteristics of Rett syndrome are very similar, each girl's needs should be looked at individually. The following are other special appliances and equipment which may be necessary: special eating utensils, protective helmet for when walking becomes unsteady, special gloves for hand-biters, bath chair or lift to be used at home, and prone board to be used as an alternate therapy position.

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Many scientists and educators, along with dedicated parents, have been working very hard in an effort to solve the puzzle of Rett syndrome. Much is known, and much more is yet to be discovered. There seems to be an understanding of the progression of the disease and the educational needs of these girls, but nothing has yet been verified about the cause of the disorder. As Dr. Hugo Moser stated at the November, 1985 conference, the most promising aspect of this disorder is that the girls were once normal. This dramatizes the real need for finding the cause and possibly a cure. Dr. Rett believes that the cognitive ability of the Rett syndrome girl is greatly masked by her physical problems and that her eyes show the great amount of understanding and sensitivity which she possesses. It seems that these are very special girls who deeply touch the hearts of everyone they meet.